Jonathan Finder Pulmonary FAQ

Pulmonology Visits

Q: When should you first see a pulmonologist (lung doctor)? How often should you be seen?

A: In general pulmonologists like to start seeing children with Duchenne by their 6th birthday and annually thereafter. Starting to see this team early help to begin to build relationships and help your child to be more comfortable with pulmonary tests and procedures. When ambulation is lost, it is important the visit frequency increase to every 6 months.

Pulmonary Function Testing

Q: What is a Pulmonary Function Test (PFT)?

A: A PFT (pulmonary function test) is a routine, outpatient test performed in the pulmonology clinic to evaluate pulmonary function. It is also called spirometry.

Q: Why is it done?

A: The PFT is a way of measuring the strength of the respiratory muscles and the breathing capacity of the lungs. This is especially important during the 2nd decade of life when the breathing muscles may begin to decline.

Q: What does it include?

A: The main test is called spirometry (literally: "measuring breath"). With simple spirometry the patient inhales (breathes in) and exhales (breathes out) fully and forcefully. The flows and volumes produced are measured and compared to a data set of a person of similar height; results are generally expressed as a percentage of predicted value.

Q: What does each component measure

A:

- FVC (forced vital capacity): the largest breath that a person can fully and forcefully breathe out (measured in liters or "L" and is reported as a percentage of a normal predicted level; "percent predicted").
- FEV_1 (Forced expiratory volume) refers to the amount of air a person can forcefully breathe out in one second. One can also measure pressures generated at the mouth with a device called a manometer.
- ~~FEV_1/FVC ratio: the volume of air breathed out forcefully in 1 second compared with the total volume of air breathed out. People with healthy lung function can blow out 80% (0.8) of their total lung capacity in the first second, so their FEV_1/FVC measurement would be 0.8.~~
- ~~FEV (Forced expiratory volume): the amount of air that the~~ person can ~~forcefully breath out (measured in liters or "L")~~
- ~~PEF (Peak expiratory flow): measures the highest pressure of the air forcefully breathed out of the lungs~~
- PEFR (Peak expiratory flow rate): measures highest flow that can be generated with effort; measured in liters per second, or L/sec, but also reported in liters per minute.

Other components include:

- End Tidal CO2: measures the level of carbon dioxide (CO2) in the last part of an exhaled breath. This is equivalent to the CO2 level in the blood.
- MEP (Maximal Expiratory Pressure): measures the maximal pressure that one can generate by exhaling
- MIP (Mean Inspiratory Pressure): measures the maximal pressure of a breath that can be generated forcefully breathing in
- PCF (Peak Cough Flow): measures the force of a cough; this is measured in liters per minute (L/min)
- SNIF: measures the force of a forceful sniff through the nose

Cough

Q: Why is cough important?

A: Until recently, pneumonia was the most common cause of hospitalization and of death in boys with Duchenne. Fortunately, with improvements in care and therapies, this is no longer the case. People with Duchenne are prone to developing pneumonia for two reasons: the main muscle of breathing IN (the diaphragm) gets weak and prevents one from getting air BEHIND mucus and secretions; the muscles of COUGHING (the abdominal muscles) get too weak to squeeze the belly and push upward on the diaphragm. Thus secretions are not cleared from the airway and can lead to increased growth of bacteria, and may eventually develop into pneumonia.

Q: How is cough measured?

A: There are several ways of assessing how effectively one can cough. The primary measurement is **peak cough flow** (PCF. We measure the highest flow rate a person can generate during a cough, by using a flow-meter (the patient coughs through the meter). This number is compared to estimates of how much flow is needed to generate an effective cough. We can also measure peak expiratory PRESSURE (rather than flow). A peak expiratory flow rate (PEFR) on a lung function test can be used as a proxy for PCF.

Q: When should cough be assisted?

A: As a rule of thumb, we recommend assisting a cough (either manually or mechanically) once the peak cough flow falls below 270 liters/minute. Once the PCF falls below 180 LPM the risk for developing pneumonia is much higher. There is no single number that is a substitute for clinical judgement. If a patient feels that he is having difficulty coughing, is clinically perceived to have a weak cough, or seems prone to pneumonia, it is very reasonable to request a device to assist with cough.

Q: What are the benefits of assisted coughing?

A: The main benefit is that with careful and aggressive assisted coughing, a patient with Duchenne will decrease his likelihood of developing of pneumonia. There are additional benefits beyond prevention of pneumonia.

- Regular (at least twice daily use) assisting of a cough will help pop open parts of the lung that might have lost volume. We all sigh and yawn to achieve this and so when people living with Duchenne lose the ability to take a full breath, they are at risk for developing areas of volume loss or collapse, which is called “atelectasis” by doctors.

- Aggressive use of a cough device will help stretch the chest wall muscles and prevent contractures of the chest wall muscles. This keeps the chest wall limber and avoids development of a stiff chest (which can interfere with coughing).

Q: What are the symptoms of ineffective cough?
A: Sadly, the development of an ineffective cough is often asymptomatic. One might not be aware that one has an ineffective cough until one gets a cold and has to cough, and finds it impossible to clear the mucus from the throat. This is why measuring lung function on a regular basis is absolutely critical: we can predict who is at risk for an ineffective cough and developing pneumonia.

Q: How is cough assisted?
A: There are 2 means of assisting cough.

- **Manually assisted cough** requires a cooperative patient and an AMBU bag to inflate the lungs. One has to fill the lungs with the bag-and-mask device and then another caregiver squeezes the chest or pushes on the abdomen to help expel air. There are videos on line demonstrating this. An AMBU bag is a relatively inexpensive device and is also light-weight.
- Since the mid-90's, **mechanically assisted coughing** has been widely available. These devices act like an old fashioned vacuum cleaner, pushing air into the lungs, then pulling the air back out again. They can be set on a timer and work automatically, can be triggered by one's own cough, or just used with the manual control of a caregiver. The effectiveness of mechanically assisted coughing is greater than that of manually assisted coughing, and is generally preferred and well tolerated. These devices are fairly expensive and not universally available**.**
- **A combination of mechanical and manual cough assistance** is superior to just mechanical or manual assistance and can be taught by your pulmonary care team.

Breathing Issues
Obstructive Sleep Apnea

People living with Duchenne have a higher rate of developing obstructive sleep apnea (brief episodes of not breathing during sleep) due to a combination of factors (low muscle tone of the upper airway, large tongue, steroid-induced obesity). Obstructive sleep apnea interferes with quality of sleep and can even lead to strain on the heart. This can occur well before the development of respiratory failure in sleep, and often occurs early in the second decade of life (mean age of 12 in one study).

Q: What is obstructive Sleep Apnea (OSA)?
A: OSA occurs when the upper airway closes off while the person is trying to breathe in, leading to the inability of air to flow into the lungs. OSA is nearly always associated with snoring.

Q: Why is treating OSA important?
A: OSA in children not living with Duchenne can lead to poor school performance and learning due to poor quality sleep, and rarely can lead to heart disease in children from the strain on the heart. Since people living with Duchenne are already very prone to heart involvement, untreated OSA can worsen cardiac disease in addition to the school and sleepiness problems as outlined before.

Q: How is OSA evaluated?
A: The gold standard for diagnosing OSA is a sleep study, which for children is performed in a pediatric sleep laboratory (patients sleep in a laboratory bed while being closely monitored).

Q: What are the symptoms of OSA?
A: Snoring is nearly universal. Excessive daytime sleepiness will occur due to poor sleep quality, and can result in falling asleep in class, in the car, in front of the TV. Grades can suffer as a result.

Q: How is OSA treated?
A: The standard treatment of OSA is use of a mask that pushes pressure into the upper airway and opens it back up. This is referred to as CPAP, which stands for "constant positive airway pressure." Because people living with Duchenne will eventually need more support than a single level of pressure, we recommend that if CPAP is needed, the device provided is later be able to deliver bilevel support (BiPAP).

Hypoventilation
Q: What is hypoventilation?
A: Hypoventilation literally means not breathing enough ("hypo" means "under" and "ventilation" means "breathing"). Inadequate breathing in sleep is common in the mid-teens. Hypoventilation leads to high blood levels of carbon dioxide, which depresses blood oxygen levels.

Q: Why is it important?
A: Hypoventilation is, in short, dangerous. It is a risk factor for developing or worsening cardiac disease and by itself increases risk of dying if not treated.

Q: How is hypoventilation diagnosed?
A: Hypoventilation is often suspected based on results of lung function testing, specifically the measure of forced vital capacity (FVC). Having a FVC less than 30% predicted is strongly associated with need for help with breathing during sleep. The diagnosis is confirmed by a sleep study in which CO2 levels are measured during sleep. Since the muscles are most relaxed during the deep portion of sleep called REM (rapid eye movement) sleep, it is critical to assess breathing during REM sleep. If the CO2 level falls during REM sleep, for example, the patient will wake up, and so will not achieve restful sleep.

Q: What are the symptoms of sleep hypoventilation?
A: Development of hypoventilation in sleep can be sneaky and silent. Symptoms often include increased frequency of awakening during sleep, nightmares of smothering or drowning, and awakening with a headache in the morning from the CO2 retention.

Q: How is sleep hypoventilation treated?
A: Treating hypoventilation during sleep requires non-invasive breathing support. The most common solution is a device called BiPAP (Respironics) or VPAP (Resmed). These devices deliver air pressure into the lungs by mask to assist one's own natural breathing efforts. The breaths delivered by the machine are timed to your own natural breathing: in other words, these devices sense the beginning of a breath and kick in some help as the patient begins to breathe in. There are many kinds of masks and other interfaces.

Q: What is the difference between BiPAP and CPAP?
A: CPAP provides a single level of pressure during breathing in and out, and is used ONLY to open the airway of a person diagnosed with obstructive sleep apnea. CPAP does NOT help with sleep hypoventilation. BiPAP provides 2 different pressures (1 to help breathe in and 1 to help breathe out), helps decrease breathing effort and helps with ventilation (getting oxygen in and CO2 out).

Q: What are the interfaces for BiPAP?
A: choices of interfaces include

- a mask that fits over the nose,
- a mask that fits over the nose and mouth,
- a mask that covers the full face and
- nasal prongs/pillows that fit just into the nose

There is always one interface that will fit best. The key here is that the device has to fit – if it is uncomfortable, it will not be used. All interfaces must be ordered by a clinician.

Q: What about hypoventilation while awake?
A: We can diagnose this stage, which always happens following the development of sleep hypoventilation, by measuring the exhaled CO2 during an office visit. Levels over 45 mm Hg are abnormal. We will prescribe daytime non-invasive BiPAP support in that setting, most commonly a portable ventilator mounted to the back of the wheelchair with a tube connected to a mouthpiece that allows the patient to pull a breath off the ventilator when needed.

Q: What are the pulmonary emergencies?
A: (1) the most common emergency is **pneumonia**. The problem with pneumonia is that the doctors may not know if the patient's low oxygen saturation is due to pneumonia or due to **inadequate breathing while awake**, so make sure that a measurement of blood CO2 (typically a finger-stick capillary blood gas) is performed.

(2) Sudden onset of shortness of breath can have many causes, such as pneumonia (above) but also can be a **mucus plug** and volume loss (**atelectasis**) from a weak cough;
(3) If shortness of breath follows a broken bone, or a severe bump, we worry about **fat embolism syndrome** (fat from the bone marrow lodging in the lung). This is a medical emergency that may be life threatening. You should go immediately to the emergency room.

Emergency Checklist:

Breathing problems

If you/your child is having problems with breathing (shortness of breath, difficulty breathing, etc.).

Remember:

- Risk: Respiratory failure. Please only give oxygen with close monitoring of CO2 levels; breathing may need to be supported (with BiPAP, for example)
- If oxygen levels are low, assisted coughing (with cough assist machine or Ambu bag) may help
- Take your equipment (cough assist, BiPAP, etc.) with you to the hospital/emergency room (ER); alert your neuromuscular team that you are going to the ER/hospital

Take the following pieces of information with you:

- Emergency card (link)
- copy of last neuromuscular note (link)
- Phone number for your neuromuscular team
    - call and let them know that you are on your way to the emergency room/hospital and why
- Anesthesia recommendations (link)
- Duchenne extubation protocol (link)
- Oxygen precautions/recommendations (link)
- Fat embolism information (link)
- take any breathing equipment that you use at home (cough assist, BiPAP, CPAP, etc.) with you
- Take any medications that you take at home with you

Broken or suspected broken bone / fall / trauma

If you suspect that you/your child may have broken a bone or has has a fall/trauma.

Remember:

- If ambulatory: Ask if internal fixation/surgery rather than casting, may be possible. Surgery may help preserve walking.
- If your child has had a fall or a leg injury, and has rapid onset shortness of breath/difficulty breathing, changes in alertness (confusion, agitation,

disorientation) this is an emergency; go immediately to the ER and alert staff that symptoms could be due to Fat Embolism Syndrome (FES).

Risk of Fat Embolism Syndrome (FES) Following a Fall /Fracture /Trauma:

- ☐ Extremely rare
- ☐ Results when fat particles enter the blood circulation, causing decreased oxygenation to the heart and/or brain
- ☐ Usually follow long bone/pelvic fractures or trauma; very rarely has occurred after orthopedic surgery
- ☐ Should be considered if the child develops shortness of breath or neurological symptoms after a fall/fracture/trauma
- ☐ See the symptoms of FES & how to care for it.

Take the following pieces of information with you:

- ☐ Emergency card
- ☐ Copy of last neuromuscular note
- ☐ Phone number for your neuromuscular team
    - Call and let them know that you are on your way to the emergency room/hospital and why
- ☐ Anesthesia/analgesia recommendations
- ☐ Oxygen recommendations
- ☐ Fat embolism information
- ☐ Take any breathing equipment that you use at home (cough assist, BiPAP, CPAP, etc.) with you
- ☐ Take any medications that you take at home with you
- ☐ Information regarding managing broken bones in Duchenne